CHILDREN'S MEDICAL RESEARCH INSTITUTE

Jeans for Genes®

2024 Year in Review

Thank you for joining the fight against childhood genetic disease.

Contents

Cover image: Joseph, Cystic Fibrosis

Welcome

On behalf of all of us at Children's Medical Research Institute—researchers, other staff, research students, and volunteers, we wish you and your loved ones a very Happy New Year.

To mark the start of 2025, we bring you this Year in Review, recapping the research highlights of the past year, for which your support has been crucial. Your dedication to Children's Medical Research Institute (CMRI) is helping our scientists to continue their world-leading efforts in the fields of cancer and genetic disorders.

Our ProCan® cancer research program now has more than 100 active national and international collaborations and efforts are already underway to translate some of our results into the clinic—initially for children's cancers. Our other main field of cancer research—the telomere and DNA repair programs—have yielded paradigm shifts in our understanding of cancer, including unravelling the mystery around how cancers respond to radiation treatment. This work could have profound effects in the future and will help many patients by improving the body's ability to activate its own immune system to fight cancer after radiation therapy.

Our expertise in proteomics has also led to significant wins in the areas of epilepsy research, blinding retinal (eye) diseases, and a life-changing discovery that may affect how Kabuki syndrome is treated. Finally, we've made major progress in our gene therapy research for a range of conditions, including inherited liver disorders where we conducted world-first gene therapy testing in whole human liver, which was widely reported in the media.

With 76 research discoveries published in world-leading journals in the past year, the successes mentioned above are just a glimpse of the breadth of our work. I hope this gives you great satisfaction, knowing so much is happening thanks to you and our community of supporters (both individuals and organisations), across Australia.

You have made a difference through your donations and fundraising involving your friends, family, coworkers and other peers, by participating in Jeans for Genes, Great Cycle Challenge, or many wonderful committee events. I am always inspired by your tireless efforts on behalf of sick children and their families. When so many of us come together, we raise the tide toward achieving CMRI's vision of finding cures for life-threatening and devastating conditions that affect more than 1 in 20 children.

Sincerely,

Roger Reddel AO
Lorimer Dods Professor & Director, CMRI

What you've helped us achieve in 2024

76
research advances published

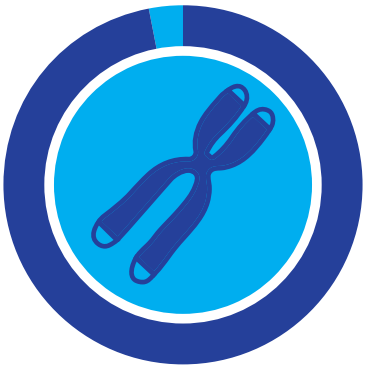
59%
of publications in top 10% of journals

737
Collaborating Institutions worldwide

Prominence* of our Research Worldwide:

Gene Therapy
(genetic diseases)

99th percentile



Telomeres
(cancer)

97th percentile

Dynamin
(epilepsy and other conditions)

88th percentile

**CMRI leads the world in these topics. For example, 99th percentile means 99% of organisations are not as recognised as CMRI in this field.*

Overall research impacts*

59% higher than the global average

*(FWCI 1.59)

Jan Pennell: a legacy of service and generosity


Jan Pennell

Jan Pennell was a force of nature—a trailblazer, a leader, and a woman with a heart as strong as her determination. Her remarkable career in the Royal Australian Air Force (RAAF), where she rose to the rank of Wing Commander, was matched by her enduring legacy of generosity: a gift in her Will to Children's Medical Research Institute (CMRI).

"Jan was truly one of a kind," shared Les, her longtime friend and executor of her Will. "She wasn't just fiercely independent—she was fiercely compassionate."

"She always believed in helping others, even if it meant pushing through her own pain."

Jan's life was one of service. After training as a nurse, she joined the RAAF and dedicated 26 years to caring for others, often during the most challenging times. She coordinated life-saving missions during disasters, including the Bali bombings in 2002, when she orchestrated the aeromedical evacuation for the Australian Air Force, saving countless lives in the critical first 24 hours.


Les, executor of Jan's Will

But Jan's kindness wasn't limited to those she met in her career. She believed deeply in the power of medical research to change lives. "Jan always said, 'We owe it to children to give them the best chance in life,'" Les recalled. It was this belief that led her to leave a gift to CMRI—a decision that will fund research breakthroughs for generations to come.

Jan's legacy extends far beyond the donation itself. It represents her unwavering belief in the importance of giving back. "She didn't have children of her own," Les explained, "but she thought of the staff she mentored and the children CMRI supports as her family."

Her life's work and her gift are a testament to her values: a dedication to protecting others, a commitment to excellence, and a fierce determination to leave the world better than she found it.

"Jan wasn't one for grand gestures or attention," Les shared, "but this gift—this incredible act of kindness—shows just how big her heart truly was. She wanted her legacy to mean something."

Through her gift to CMRI, Jan has ensured that her compassion will ripple through the lives of countless families. Her generosity gives hope to parents facing the unimaginable and ensures that the researchers at CMRI can continue their groundbreaking work to cure childhood diseases.

Jan's story reminds us all of the power of leaving a gift in our Wills. Like Jan, we each have the opportunity to create a legacy that lasts beyond our time. A legacy of hope. A legacy of change. A legacy that gives every child the chance to live a healthy, happy life.

Team CMRI

Great Cycle Challenge: pedalling towards a cancer-free future

Every year thousands of dedicated bike riders take part in the Great Cycle Challenge to raise money for our research, but this year there was a new kid on the block—one of Australia's top cancer researchers, CMRI's Director, Professor Roger Reddel.

Since 2013, riders have raised more than $40 million to help our scientists work on personalised approaches to cancer diagnosis and treatment.

Scientists like Professor Reddel want to change the lives of kids like Annabelle.

The gorgeous three-year-old came home from preschool one day with a lump on her head after a fall. Her parents, Mel and Brendan, weren't concerned until it developed into a sore, and then suddenly, she came out in bruises all over her legs.

"When our doctor saw her bruises, he said, 'I'm sorry I hope I'm wrong, but that is a telltale sign of leukaemia'."

Brendan said they were just shellshocked, and within 24 hours she was having a lumbar puncture (also known as a spinal tap), and had started chemotherapy.

"Even then, I just kept thinking that someone was going to tell us they got it wrong," he said.

The good news for the family was they had caught the cancer very early. Since then, Annabelle has been taking oral chemotherapy tablets at home, receiving weekly chemotherapy at the hospital, and is soon starting a new clinical trial, which her family has been told may increase her survival chances by an additional 10%.

"I look at the timeline of how fast Annabelle was diagnosed and treated and it's amazing," Brendan said. "We still have a long road ahead of us, but it's exciting that she's been offered this new treatment that improves her chances—and that's because of research."

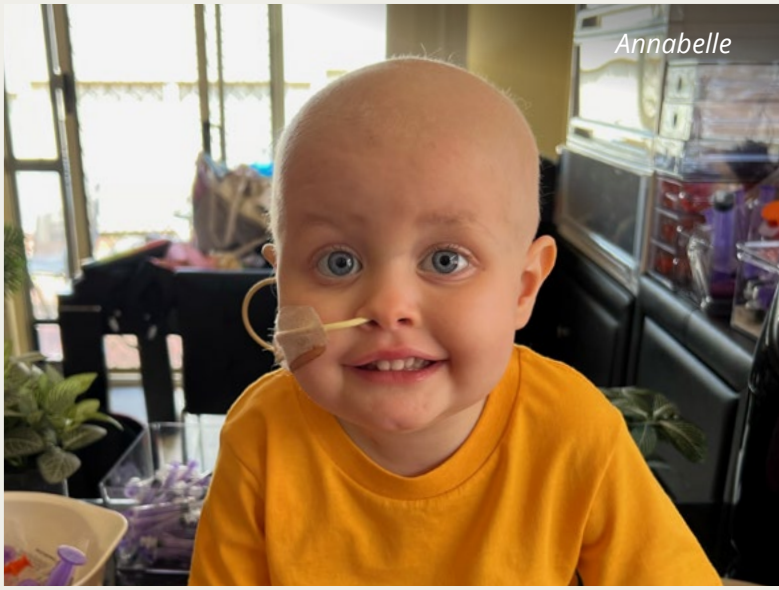
This year, Professor Reddel decided to jump on a bike himself, as part of a team of scientists and staff at CMRI who took part to fundraise, but also to say thank you to all of our supporters.

"I'm not usually a bike rider, but the Great Cycle Challenge is such an important event that I decided this year to find a way to join the action by riding a stationary bike," he said.

"GCC is an incredibly important boost to the cancer research being done at CMRI. The opportunities to make big advances have never been greater. The greater the funding, the faster we will make significant progress against this disease," he said.

Roger Reddel

Annabelle and her dad, Brendan

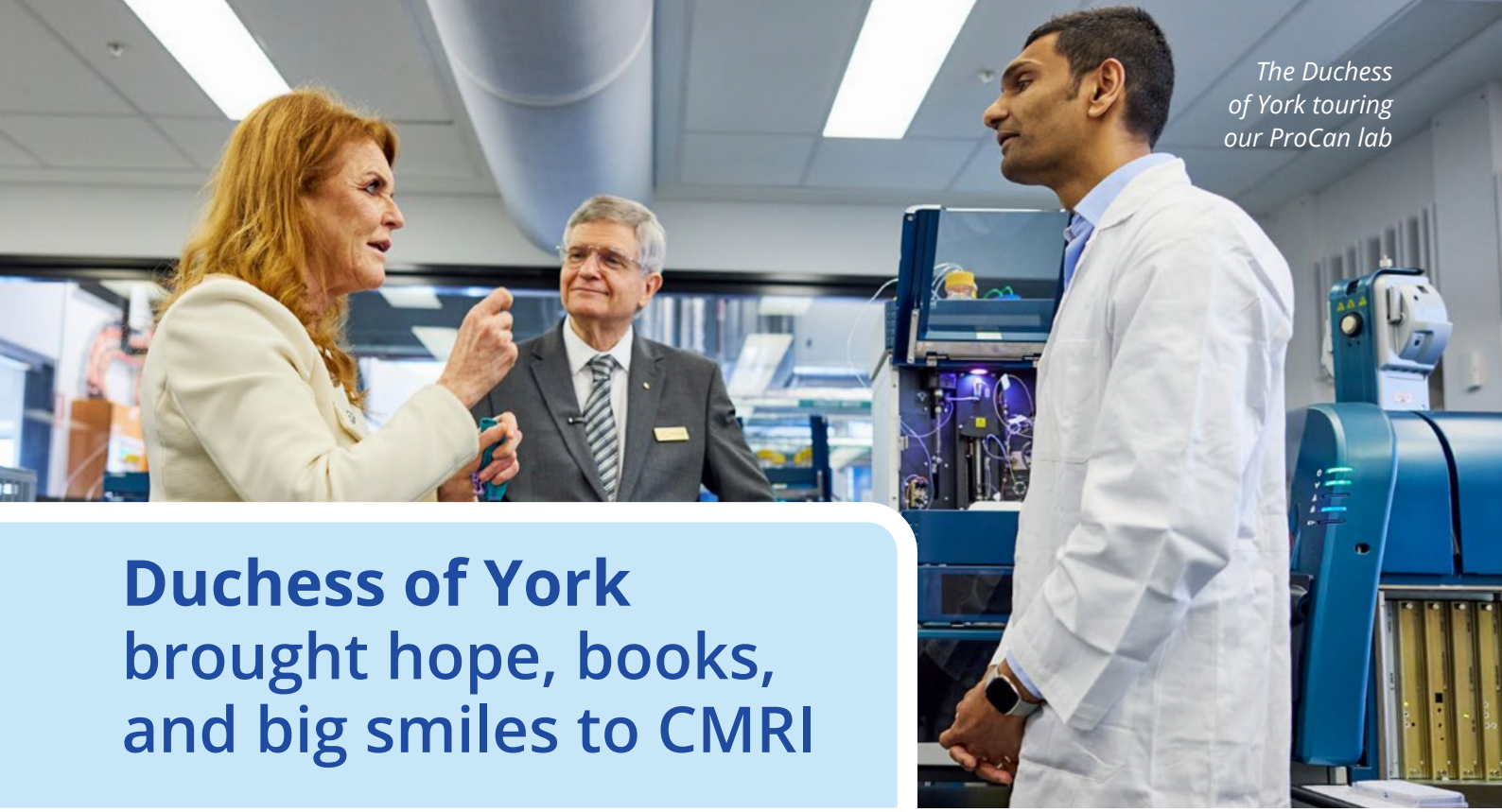


Annabelle

Margaret's epic ride against cancer!

At nearly 80, Margaret Rowlings is an unstoppable force. A breast cancer survivor, Margaret began cycling in 2013 to stay active through her treatment, sparking a journey that led her to become a Great Cycle Challenge (GCC) Champion. This year marks her seventh-year riding to support CMRI's fight against childhood cancer, and she's raised nearly $12,000 with the backing of her Kyogle community.

Margaret recently visited CMRI and was deeply moved by the groundbreaking work of ProCan. Her journey has even earned her the title of Kyogle Sports Person of the Year, and her GCC page is filled with heartfelt messages to her supporters. From survivor to community hero, Margaret's dedication proves that age is just a number when it comes to making a difference.



The Duchess of York touring our ProCan lab

Duchess of York brought hope, books, and big smiles to CMRI

It was a unique privilege for everyone at Children's Medical Research Institute to welcome Sarah, Duchess of York, into our labs, and to have her advocate for our cancer research during an Australian book tour.

The Duchess was visiting our shores to promote her new children's book *Flora and Fern: Wonder in the Woods* which is being sold at Dymocks stores across Australia. The Duchess did a special book signing hosted by Jeans for Genes partner, Seed Heritage with all profits from the book sales that day going to CMRI.

After meeting our Board members, The Duchess then went into ProCan where Professor Roger Reddel explained how the project is hoping to change the way cancer is diagnosed and treated.

The Duchess also met some Jeans for Genes families and gave them copies of her books.

"CMRI is giving hope to children. I've just been diagnosed, with breast cancer. I know that feeling of fear. I know that feeling when you think there are no solutions," The Duchess said.

"People need to know there is a place like ProCan that is researching to find health solutions. That's why I've come to CMRI, to ProCan, to say to everybody out there—you've got this, we got you, we're going to help you."

The Duchess meets our kids and staff

The Duchess used her own diagnosis to implore others to support research.

"The drive from the hospital was the longest drive I've ever had. Because you're in abject fear of your own demise. I decided to go for a very quick mastectomy because I didn't want it to spread any more.

"Jane, my sister, and I lost our father to cancer. I lost my stepfather to cancer. I lost my grandmother to cancer. Cancer is obviously very much in the forefront of our minds.

"I think ProCan is listening, I think Roger is listening. He said, 'I would want to know that if a person in my family has cancer there are solutions out there.' Roger and ProCan are incredible."

Skippers Zian, Kaden and Ayaan

Skipping for good: the Aluwihare family's inspiring journey

We had a record number of entrants in 2024 for our 100 Skips a Day Challenge, but for one family of skippers it was all about giving back.

In 2024, the skippers in our loyal community raised $320,000—the largest amount since the challenge was launched three years ago.

One of the biggest fundraisers was the Aluwihare family, including 12-year-old twins Zian and Kaden and their 10-year-old brother, Ayaan.

Mum Ruby said from a young age the family encouraged the boys to think of others, particularly because their cousin lives with a genetic condition.

"I loved skipping with my kids, as it gave us some quality fun family time together each day, often resulting in us falling over the floor laughing at ourselves," Ruby said.

"There were times the kids were tired, but the encouragement they received from their friends, family, and school community was incredible. They even took their skipping ropes with them when they went skiing!"

Ruby said the private Facebook group, where skippers shared videos of themselves completing their daily skipping challenge, provided an amazing community feeling.

"It was lovely to feel part of an inclusive, welcoming group. Somewhere we all came together to encourage and cheer on each other."

As the boys' parents both work in the medical field, Ruby said they valued the research being done by the scientists at CMRI.

"Medical research is something we are very familiar with," she said. "It helps us identify new, evidence-based and improved ways to optimise health outcomes. I am personally so grateful that we live in a country where we have access to health and medical care."

"Giving back is a part of my culture. As a parent, I strive to give my boys a firsthand account of how and why we should show generosity to others. As they grow into young adults, I hope that they go out and start a ripple effect of generosity and kindness in the world. Proactively giving back helps my boys develop a strong value system.

"I'd like to take the opportunity to thank all those involved with Jeans for Genes Day in doing the work to put this challenge together.

"In my son's words, 'This is the BEST feeling, helping sick kids makes me happy, warm and fuzzy in my tummy—like a big cuddle.'"

Joseph and parents, Sarah and Carmello, meet staff at Jeanswest

Fashioning a future for medical research

One of the highlights of 2024 was the launch of our new Retail Alliance, which saw major Australian retailers band together to raise hundreds of thousands of dollars for our research through Jeans for Genes.

The concept of the Retail Alliance was simple—getting some of the biggest names in Australian fashion to pledge their support for our research. This meant collecting donations in their stores, having promotions where the stores gave a percentage of sales to CMRI, and encouraging staff to promote the Alliance.

Jeanswest was the premier partner leading the charge under General Manager Anne Natale.

"Retailers have been doing it tough, but these kids have it tougher," said Anne.

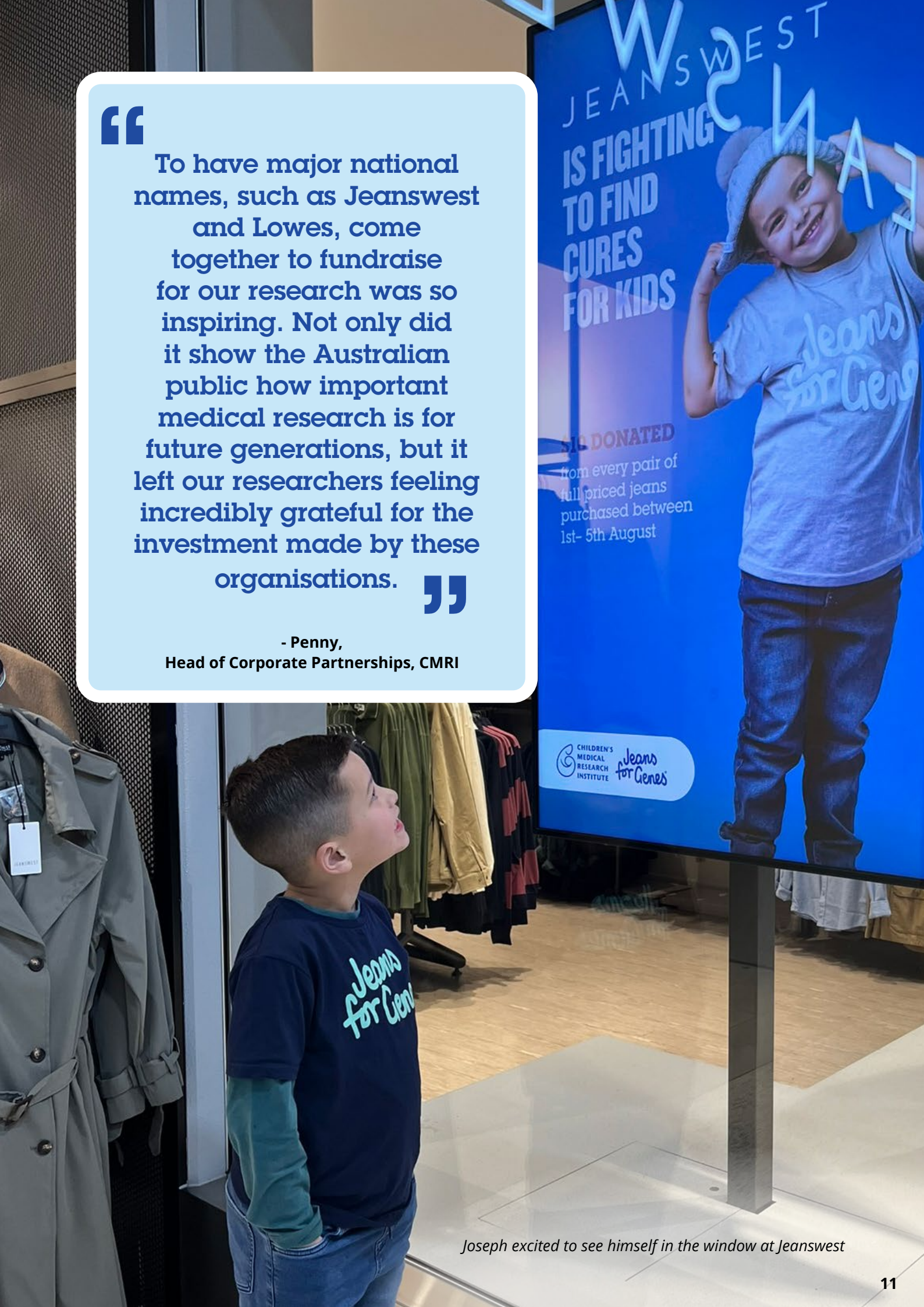
"When you think about what the kids and their families are going through, it feels good to do whatever we can to help. It inspires my staff and keeps us all going.

"It's when things are hard that you need to band together and help one another."

Lowes also joined the Retail Alliance as another top tier partner. A long-time supporter of Children's Medical Research Institute, Lowes owner, Linda Penn, says: "We've supported this vital cause since 1998, and when we heard about the Retail Alliance, we wanted to be a big part of it."

Other top retailers who took part were Seed Heritage, Commonry, and Unison.

The Retail Alliance also recieved support from Dave String Advertising!, Kepler Analytics, Ultimate Edge Communications, and Shopper Media.



> To have major national names, such as Jeanswest and Lowes, come together to fundraise for our research was so inspiring. Not only did it show the Australian public how important medical research is for future generations, but it left our researchers feeling incredibly grateful for the investment made by these organisations.

- Penny,
Head of Corporate Partnerships, CMRI

Joseph excited to see himself in the window at Jeanswest

Professor Ian Alexander

Dr Spela Mirosevic

Rare disease day: hope, connection, and purpose

The impact of our research is always far-reaching, but one mum said she hoped our first ever Rare Disease Family Day gave scientists a true sense of purpose in their work.

Families, researchers, and clinicians came together for the event to talk specifically about the progress of gene therapy programs at CMRI.

Information was shared between CMRI scientists researching the latest in gene therapy technologies, clinicians from the Sydney Children's Hospital Network (SCHN) who run clinical trials for gene therapy, and families from across Australia whose children are living with rare and ultra-rare serious genetic conditions.

A special guest of the event was Slovenian mother Dr Spela Mirosevic who, together with her husband Samo, started the international CTNNB1 Foundation after her son Urban was diagnosed with this rare condition. Many children with this condition cannot walk or talk.

"In the ideal world, you would go to the doctor, who would give you the diagnosis and then administer the treatment," Dr Mirosevic said.

"Unfortunately, we do not live in that ideal world...at least not yet. However, we live in a time where there is hope with gene therapy."

Dr Mirosevic reached out to CMRI based on recommendations from families and rare disease experts and has been working for the last three years with Professor Leszek Lisowski, leader of CMRI's Translational Vectorology Research Unit, on a novel experimental gene therapy for CTNNB1.

Australian mother, Lucy Mort, attended with her son Teddy who has CTNNB1.

"Rare Disease Day gave us an amazing opportunity to meet other families like ours and allowed us to hear from incredible researchers and medical professionals who truly care about our children and our situation," she said.

"Learning about the intersection between science and medicine and how we can take research into the field to make a meaningful impact in the lives of patients was very enlightening. We learnt about the complexity of clinical trials, especially in the context of gene therapy, but it gave us hope for the possibilities of gene therapy for our child."

Professor Leszek Lisowski is always happy to open his lab to families

> **Researchers and families staying connected is mutually beneficial. For families, we get to learn more about our child's condition from a scientific standpoint, whilst gaining a sense of hope for our child's potential treatments. I think scientists benefit from family connections because they gain a true sense of purpose in their work and understand why their daily efforts are so incredibly valuable.**
>
> **- Lucy, Teddy's mum**

Sophia's triumph: from survivor to big sister

Sophia adores being a big sister to her two brothers

She may only be four years old, but Sophia is very aware that she was once extremely sick with cancer.

"She's definitely aware," mum Elizabeth said. "She's actually come to be quite proud of the scar from her central line."

Sophia was a face of Jeans for Genes in 2022. She was only a year old when she developed swelling around her eye which led to the diagnosis of a germ cell tumour by oncologist and CMRI Board member, Dr Luciano Dalla Pozza. He told the family that the treatment plan had a 70-80% success rate.

Sophia underwent chemotherapy and surgeries and is now three years in remission.

The family agreed to be part of Jeans for Genes because they knew that research was the key to Sophia getting the right treatment.

"Three years is very significant," Elizabeth said. "We've been told it would be very rare if her cancer came back now."

Sophia with brother Hamish

Sophia and Hamish

Sophia, Hamish and Luke

Today she is a bright and bubbly child. Her only remaining impact, outside of the scars, is loss of 80% vision in one eye. In general, she is strong and healthy.

"She loves preschool, she loves, painting and she really loves butterflies," Elizabeth said.

One of her biggest achievements since her diagnosis was becoming a big sister to brothers Hamish and Luke.

"She is the best sister, she's so good with them. She's so caring and so patient," her mum says.

While Sophia doesn't enjoy the blood tests that are part of her regular check-ups, Elizabeth said there is an awareness and appreciation of what she's been through.

"She comments that she's so lucky the doctors gave her the medicine that made her better."

Amity's big fancy fundraiser

Amity prepares to draw the raffle winners at her fundraiser

It was a challenging year for so many but, as always, the incredible CMRI community rallied around Jeans for Genes in 2024, raising more than $2 million.

All thanks to those of you who took part in Bake it Blue, wore your jeans to work and school, or helped make our 100 Skips a Day more popular than ever!

We are also heartened by the incredible ways that families who live with genetic diseases fundraise for us—like Amity's.

When Amity was chosen to become a face of Jeans for Genes her family knew they wanted to make it a big deal, so they organised "Amity's Big Fancy Fundraising Event".

The Jeans for Genes event was generously hosted by El Patron Restaurant in Gregory Hills in Sydney and attended by more than 100 members of Amity's family.

"Having Amity as a face of Jeans for Genes has allowed us to educate our friends and family about the impact of living with genetic disease," mum Cass said.

Amity and family visit Lowes


Amity loved visiting the CMRI labs

"We also saw it as the perfect opportunity to fundraise for research.

"When you have a child with a rare condition you can feel very alone, especially when there isn't much research happening in that area. Being a part of Jeans for Genes and fundraising for CMRI meant that we could put our energy into something that may help kids like Amity one day—so scientists can learn more about rare diseases."

Jeans for Genes Campaign Manager, Greg Noel-Butterworth said, "We are so grateful for all the support that the public have shown us this year. Whether they donated money or their time to help us fundraise, it is greatly appreciated.

"Most importantly our scientists would like to thank each and every person who has donated to their research."

"Many wouldn't be able to do their PhD research, finish their major project, or complete the next step towards an amazing discovery if it wasn't for the Australian public. Thank you from all of us and from the children who will benefit from their work."

Unlocking the secrets of cancer treatment

One of the biggest mysteries in cancer research was solved by a CMRI team at the end of 2024—which will open new opportunities to improve treatment and increase cure rates.

The findings were published in *Nature Cell Biology* by first author Dr Radoslaw Szmyd of CMRI's Genome Integrity Unit, which is led by Professor Tony Cesare.

Radiation therapy (also called radiotherapy) is a critically important type of cancer treatment. Scientists have struggled for decades to understand why radiation therapy kills cells from the same tumour in different ways.

This is important to understand, because some forms of cell death are unnoticed by the immune system, while others trigger an immune response that kills other cancer cells.

Unleashing the patient's immune system to kill cancer cells and clear tumours is a major goal of cancer treatment.

"The surprising result of our research is that DNA repair, which normally protects healthy cells, determines how cancer cells die following radiotherapy," said Prof Cesare. "The DNA inside our cells is constantly experiencing damage, and DNA repair is happening all the time to fix that damage and keep our cells healthy. Now, however, it seems these repair processes can recognise when overwhelming damage has occurred and instruct a cancer cell how to die."

Professor Tony Cesare

Dr Radoslaw Szmyd

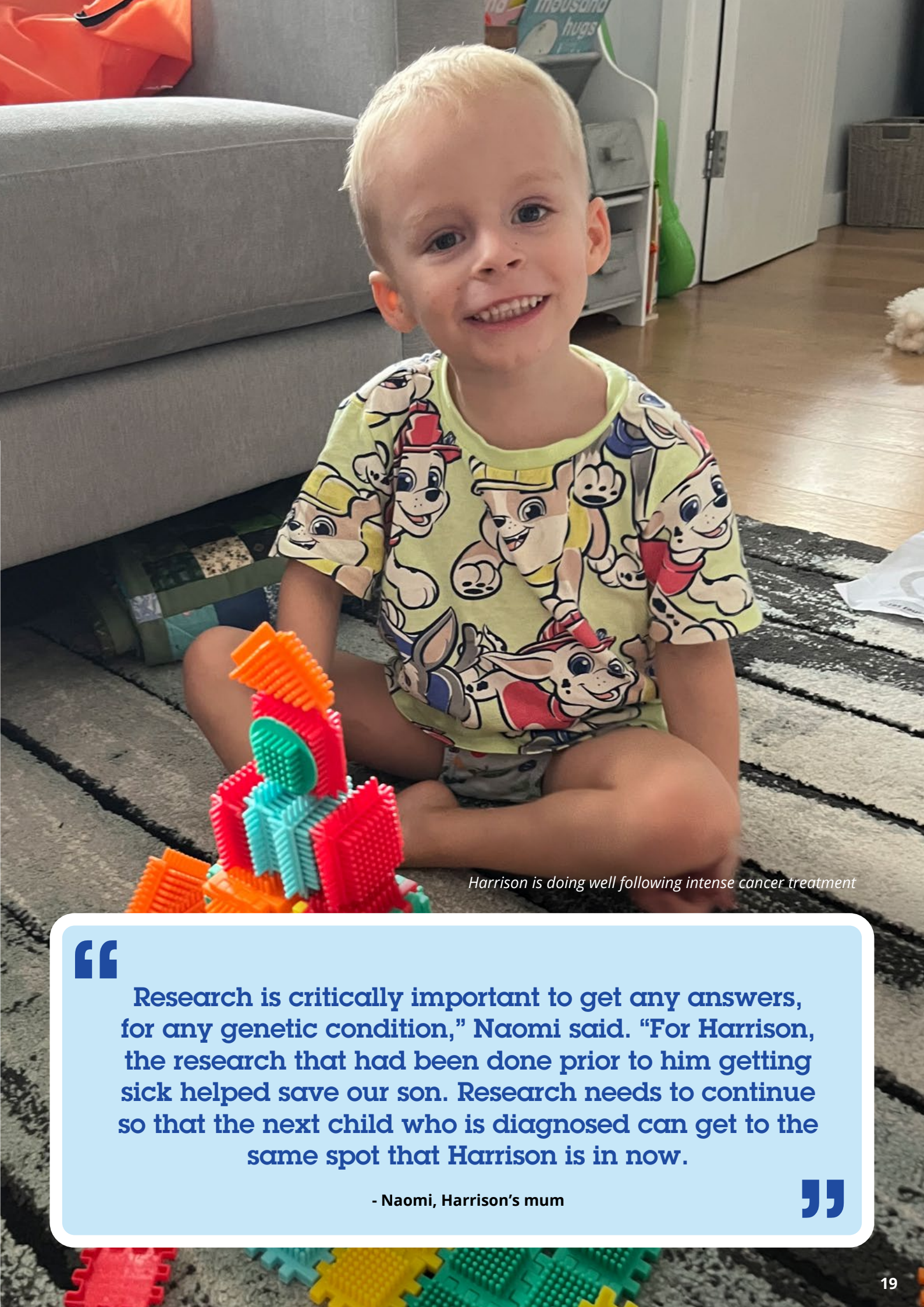
Co-project lead, A/Prof Harriet Gee, a radiation oncologist from the Western Sydney Local Health District Radiation Oncology Network, said these findings answer a clinical question that has puzzled the field for 30 years.

"We have now found that the manner in which tumour cells die after radiotherapy depends on the engagement of specific DNA repair pathways, particularly when radiation is given at very high, focused doses. This opens new opportunities to enhance radiation efficacy through combination with other therapies, particularly immunotherapy, to increase cancer cure rates."

Prof Cesare shared that Dr Szmyd dedicated six years to solving this "incredibly difficult nut to crack". He added, "The perseverance needed for a project like this speaks volumes about Radek and the team.

"We all know someone touched by cancer, so finding something with the potential to truly improve lives is incredibly rewarding."

Jeans for Genes ambassador, Harrison, went through very intense cancer treatment over a fairly short period of just six weeks. Mum Naomi said that research, like that done by Prof Cesare, is vital to help improve treatments and save more children's lives.



Harrison is doing well following intense cancer treatment

> Research is critically important to get any answers, for any genetic condition," Naomi said. "For Harrison, the research that had been done prior to him getting sick helped save our son. Research needs to continue so that the next child who is diagnosed can get to the same spot that Harrison is in now.
>
> **- Naomi, Harrison's mum**

Arato (Jeans for Genes ambassador, second left) and Jenna (Paralympian, right) were among guests at the Inherited Retinal Disease Patient & Family Engagement Day

A visionary year for stem cell innovation

Associate Professor Anai Gonzalez Cordero

2024 was an extraordinarily significant year for our Stem Cell Medicine Group who were involved in several State Government initiatives to progress research forward in this area.

Group leader, Associate Professor Anai Gonzalez Cordero, started the year with an invitation to become a founding member and launch the NSW Government's Organoid Innovation Centre—a first of its kind in Australia.

Soon after this, A/Prof Gonzalez Cordero was asked to be part of a NSW Health and Investment NSW led delegation of eminent scientists showcasing the State's contribution to scientific innovation in Australia, at the BIO24 event in San Diego, USA.

"Such events on an international platform are important to place Australia at the forefront of new biotechnologies like our stem cell and organoid products," says A/Prof Gonzalez Cordero.

"The technology is here, but companies need to start embracing it, and there will be an education period where the old established methodologies are replaced. It demonstrates that there is investment and state-of-the-art research happening here in NSW."

The Stem Cell Medicine Group was also involved in organising an Inherited Retinal Disease Patient and Family Engagement Day in Sydney, with attendees from all over Australia and New Zealand.

The aim of the day was to educate patients and their families about the latest technology being used in the research of retinal diseases, such as stem cell medicine, and was attended by media personalities and international sporting identities living with vision loss.

Finally, the year ended with the group becoming involved in the NSW Government's newly formed Non-Animal Technologies Network (NAT-Net).

The aim of the Network is to develop resources for medical researchers and to work towards technologies that reduce the need to use animals when testing potentially life-saving therapies.

A/Prof Gonzalez Cordero said it was fantastic to be a founding member.

"This is just the beginning of a very exciting time. It's important to emphasise this initiative will connect researchers across Australia and internationally."

> Lots of therapies fail in clinical trials because the testing is not optimal. We are working to enhance organoids so that they offer better predictive models for disease.
>
> **- A/Prof Gonzalez Cordero**

CMRI's Dr Eden Robertson, A/Prof Anai Gonzalez Cordero and Prof Robyn Jamieson at the launch of the Inherited Retinal Diseases Research Priorities report

Advancing eye genetics for a brighter future

Professor Robyn Jamieson

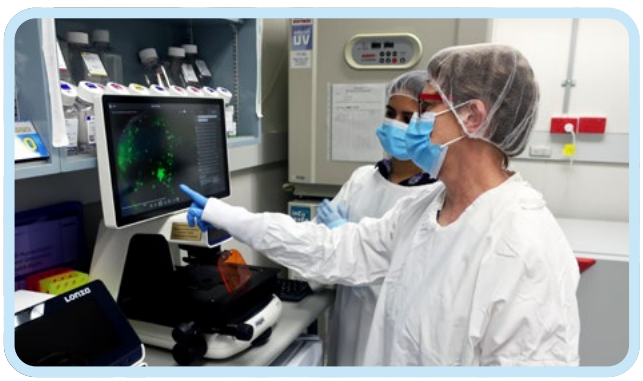
The work being done in our labs at Children's Medical Research Institute is reaching further into the community, as evidenced by our Eye Genetics team being involved in a new analysis of the impacts of Inherited Retinal Disease on patients like Paralympic swimmer, Jenna Jones.

In 2024, this project was awarded a National Health and Medical Research Council (NHMRC) Partnerships Projects grant.

Professor Robyn Jamieson is Head of the Eye Genetics Research Unit at CMRI. She is also Head of the Eye Genetics Clinic at The Children's Hospital at Westmead, has affiliations with Save Sight Institute (SSI) and is Head of Genomic Medicine at the University of Sydney.

Prof Jamieson says, "Understanding the impact of genetic variants on patients and families and developing and delivering therapies takes time.

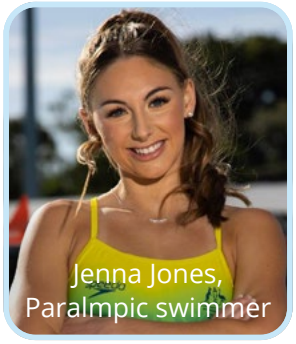
"This project will allow us to gather information about the impacts of inherited retinal diseases (IRDs) on patients, enabling improved diagnoses and delivery of, and access to, targeted therapies."



The collaborative project aimed to estimate the national social and economic impacts of specific IRDs and the cost effectiveness of using genomic and functional genomic investigations and precision medicine. This will support translating genetic diagnosis and targeted therapies into clinical practice and policy, thereby improving the health and life of patients with IRDs and their carers. The CMRI, SCHN and SSI Eye Genetics Research team work closely with investigators from Macquarie University's GenIMPACT, University of WA, University of Melbourne, and University of Auckland.

Access to targeted therapies and development of these therapies is contingent on a molecular diagnosis through genomic and functional genomic testing, where funding for these investigations is not always available.

Paralympic swimmer, Jenna Jones, is a patient of Professor Jamieson's and also an ambassador for the work done at CMRI through Jeans for Genes. She lives with degenerative vision impairment known as cone-rod dystrophy.



Jenna Jones, Paralmpic swimmer

"What I want to get across to people is that everyone's independence looks different," says Jenna. "My diagnosis was so important, and the research that made that happen is amazing. Knowledge is power."

Pioneering gene therapy solutions

Associate Professor Grant Logan

When it comes to solving complex problems, CMRI has the best of the best. This was proven when Associate Professor Grant Logan won a highly competitive NSW Government research grant for innovation.

A/Prof Logan in CMRI's Gene Therapy Research Unit received funding from the NSW Office for Health and Medical Research for his work to develop a new strategy that will enable greater patient access to life-saving gene therapies.

Gene therapy has proven to be life-changing for many patients who otherwise have few or no treatment options.

Infants diagnosed with the deadly disease, spinal muscular astrophy (SMA) are one example of how profoundly gene therapy can change a child's life.

SMA is an inherited neuromuscular disorder where children born seemingly healthy gradually lose control of their muscle function and eventually even lose their ability to breathe; in the most severe cases, the children die within the first two years of life. Gene therapy is changing this outcome with a single injection—the earliest treated newborns are now reaching their seventh birthdays and meeting all their growth milestones.

The most efficient gene delivery systems for this task are vectors based on viruses, and the current SMA gene therapy uses a harmless virus named adeno-associated virus (AAV) to carry genetic information into human cells. Unfortunately, some infants are not eligible for the gene therapy because their body has natural immunity to the AAV vector.

"Some children and adults have pre-existing immunity to the AAV viral coat protein, from natural exposure to similar viruses circulating in the community," he said.

"This is a major obstacle to treatment, because it means a proportion of patients cannot receive life-saving AAV-based gene therapies.

"We plan to tackle this challenge head-on by developing a way to 'cloak' the AAV, so it can successfully enter target tissue to deliver gene therapy."

Alessia is living proof of the life-saving impact of research—she was the first child in Australia to receive the new gene therapy for SMA and is now a thriving six-year-old.

Her mum Adriana and dad Adam visited the Gene Therapy Research Unit and were amazed by the work being done.

"When we went into CMRI and we saw the way people were fawning over her in delight at seeing first-hand how gene therapies that they are working on can save lives—it really makes you step back and marvel," Adriana said.

"Not only has this changed our lives, but it also changed the entire course of the nature of this disease from being a death sentence less than 10 years ago to now one of three gene therapy treatments on the Pharmaceutical Benefits Scheme. This is life changing and unimaginable just a few years ago. Now you look at Alessia and you have no idea she has this deadly condition."

Gerringong Committee members, (Left to right) Gwen Wishart, Sue Dmetreson

Fundraising with heart: committees making a difference

It was another extraordinary year for our amazing Fundraising Committees who continue to impress us with their unique ways to raise funds to support research, from ferret racing to high teas, bingo, and quiz nights.

Strathfield Committee held a quiz night, a morning tea, and a gala dinner with one of the faces of Jeans for Genes, Joseph, and his family as special guests.

Hills Committee held their hugely popular Mothers' luncheon and a Melbourne Cup luncheon.

Gerringong Committee celebrated Mother's Day with a high tea, as well as their annual Quilt and Craft Show, and a Melbourne Cup luncheon.

Canberra Committee invited guests to the National Museum of Australia for a very delicious Jeans for Genes high tea.

In the small regional town of Quirindi, the whole community came together for a Long White Lunch, which was a huge success.

It was all about the fun of ferret racing for the Goulburn and Surrounds Committee who continue to attract a good crowd—including many scientists—for their unique event.

Judith Hyam Committee held their first Denim and Diamonds trivia night, Maroota Committee continued their wonderful fashion show and bingo night, while Northern Beaches once again had a fantastic games day.

One of the longest running and biggest events is the Wagga Wagga Christmas Fair which continues from strength to strength.

Mudgee Committee

Judith Hyam Committee members
Trish Friend, Nola Barrie, Rhoda Booth.

Get involved!

We rely on the generosity and dedication of people like you to continue fighting against childhood diseases. Here are some of the many ways you can join the team!

Volunteer
with us

Leave a
gift in your Will

Fundraise

Sign up to become a
research champion

Donate now
cmrijeansforgenes.org.au

Amity, 5
CM-AVM2

Amity's condition is so rare the future is unknown. Her family dreads each brain scan, each test, but they hope research will show the way through.

Gem loves unicorns and mermaids, but her family know that real magic lies in gene therapy for rare genetic eye conditions.

Gem, 8
Genetic Blindness

Joseph, 6
Cystic Fibrosis

Under Joseph's cheeky smile is a child kept alive by medication and his parents' hope that one day research will lead to a cure.

Jon wasn't supposed to live past his first birthday, but gene therapy changed his family's life forever.

Jon, 5
Spinal Muscular Atrophy

Once again a huge

Thank You

from the team at CMRI.

We look forward
to even more
successes in 2025.

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